ABSTRACT

The present invention is based on sequencing genomic DNA from human chromosome 6 and cDNAs to define the genomic structure of estrogen receptor alpha genes and novel polymorphism/haplotypes in the estrogen receptor gene/protein. Such polymorphism/haplotypes can lead to a variety of disorders that are mediated/modulated by a variant estrogen receptor, such as a susceptibility to cancer, osteoporosis, cardiovascular disorder, etc. Based on this sequencing approach, the present invention provides genomic nucleotide sequences, cDNA sequences, amino acid sequences and sequence polymorphism/haplotypes in the ESR-alpha genes, methods of detecting these sequences/polymorphism/haplotypes in a sample, methods of determining a risk of having or developing a disorder mediated by a variant estrogen receptor and methods of screening for compounds used to treat disorders mediated by a variant estrogen receptor.